

Note: The following information is provided by the author(s) and has not been reviewed by *GeneReviews* staff.

Table 3. Pathologic Allelic Variants

Gene	Exon/Intron	Mutation	Allelic Percentage	Protein Mutation
<i>EIF2B1</i>	Intron 2	IVS2+1G>A	<1%	S84ins22aa, stop
	Exon 6	A622T	<1%	N208Y
	Exon 7	A622T		N208Y
<i>EIF2B2</i>	Exon 4	C512T	<1%	S171F
	Exon 4	C547T	<1%	R183fs
	Exon 4	548Gdel	<1%	R183fs
	Exon 4	C586T		P196S
	Exon 5	G599T	<1%	G200V
	Exon 5	G599C	<1%	G200A
	Exon 5	607-612delinsTG	<1%	M203fs
	Exon 5	A638G	8.2%	E213G
	Exon 6	A818G	<1%	K273R
	Exon 7	C871T		P291S
	Exon 8	G910T	<1%	E304X
	Exon 8	T947A	<1%	V316D
	Exon 8	G986T	<1%	G329V
<i>EIF2B3</i>	Exon 1	G136A	<1%	V46I
	Exon 2	C260T	<1%	A87V
	Exon 3	A407C	<1%	Q136P
	Exon 6	G674A	<1%	R225W
	Exon 8	T1023G	<1%	H341Q
	Exon 9	1193-1195delTG	<1%	V398fs
<i>EIF2B4</i>	Exon 7	C683T	<1%	A228V

Gene	Exon/Intron	Mutation	Allelic Percentage	Protein Mutation
<i>EIF2B5</i>	Exon 7	G626A	1.5%	R209Q
	Exon 7	C625T	<1%	R209X
	Exon 8	C728T	3%	P243L
	Exon 9	T806G		L269R
	Exon 11	C1069T		R357W
	Exon 11	G1070A		R357Q
	Exon 11	G1091A		R364Q
	Exon 11	C1120T	1.1%	R374C
	Exon 11	C1172A	<1%	A391D
	Intron 11	IVS11+1G>A	<1%	E397ins11aa
	Exon 13	T1393C	<1%	C465R
	Exon 13	C1447T	<1%	R483W
	Exon 13	T1465C	1.1%	Y489H
	Exon 1	C47A	<1%	A16D
<i>EIF2B5</i>	Exon 1	G161C	<1%	R54P
	Exon 1	T166G	<1%	F56V
	Exon 2	T203C		L68S
	Exon 2	T218G	1.5%	V73G
	Exon 2	G220A		A74T
	Exon 2	G241A	<1%	E81K
	Exon 2	A271G	7.1%	T91A
	Exon 2	A318T	<1%	L106F
	Exon 3	G338A	27.4%	R113H
	Exon 3	C337T	<1%	R113C
	Exon 3	C406T	<1%	R136C
	Exon 3	453-454del		Y152fsX12
	Exon 4	C545T	<1%	T182M
	Exon 4	G584A	2.2%	R195H

Gene	Exon/Intron	Mutation	Allelic Percentage	Protein Mutation
	Exon 4	C583T	<1%	R195C
	Exon 4	G592A	<1%	E198K
	Exon 6	792delTinsACA	<1%	F264fs
	Exon 6	G806T	<1%	R269L
	Exon 6	C805G		R269G
	Exon 6	G806A		R269Q
	Exon 7	G896A	<1%	R299H
	Exon 7	G925C	1.5%	V309L
	Exon 7	G929T		C310F
	Exon 7	C943G	<1%	R315G
	Exon 7	G944A	<1%	R315H
	Exon 7	C943T	<1%	R315C
	Exon 7	C967T	<1%	P323S
	Exon 7	T1003C		C335R
	Exon 7	C1015T	3.4%	R339W
	Exon 7	G1016C	<1%	R339P
	Exon 7	G1016A	<1%	R339Q
	Exon 7	G1016T		R339P
	Exon 7	A1028G	<1%	Y343C
	Exon 7	A1153G	<1%	I385V
	Exon 8	G1157T	<1%	G386V
	Exon 8	A1160G	<1%	D387G
	Exon 8	C1264T	<1%	R422X
	Exon 8	T1274G	<1%	L425R
	Exon 8	C1280T	<1%	P427L
	Exon 8	T1289C	<1%	V430A
	Exon 9	C1340T	<1%	S447L
	Exon 9	G1444ins17	<1%	G481fs493X

Gene	Exon/Intron	Mutation	Allelic Percentage	Protein Mutation
	Exon 9	G1459A		E487K
	Exon 10	A1484G	<1%	Y495C
	Exon 13	1813delC		L605fs
	Exon 14	T1882C	<1%	W628R
	Exon 14	G1884A	<1%	W628X
	Exon 14	G1948A	1.2%	E650K
	Exon 14	1996del21	<1%	664del7

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Mutations can affect all regions of the five eIF2B subunits. From Leegwater et al 2001; van der Knaap et al 2002; van der Knaap et al 2003 ; Fogli, Schiffmann, Hugendubler et al 2004; Ohtake et al 2004; Ohlenbush et al 2005; Pronk et al 2006; Scali et al 2006; Fogli et al 2007; Matsui et al 2007.